Phenotypic Expression in M.3243A>G Carriers Is Complex

Abstract:

Keywords: mtDNA, point mutation, cardiac involvement, cardiomyopathy, multisystem, mitochondrial.

Letter to the Editor

With interest we read the article by Chau et al., 2020 about a 57yo female with a mitochondrial disorder (MID) due to variant m.3243A>G, manifesting with hypoacusis, colonic pseudo-obstruction, bilateral basal-ganglia calcification, and heart failure due to hypertrophic cardiomyopathy (hCMP) (Chau, E. M. et al., 2020). We have the following concerns.

We do not agree with the statement that “mitochondrial cardiomyopathy usually presents in childhood” (Chau, E. M. et al., 2020). There are a number of MID cases, in which CMP develops not earlier than in adulthood (Niedermayr, K. et al., 2018).

A shortcoming of the study is that echocardiography was not reviewed for left ventricular hypertrobrubeculation/noncompaction (LVHT). LVHT is a myocardial abnormality of the apex, characterised by hypertrabeculation of the inner myocardial layer distal to the papillary muscles, which is most frequently associated with MID and can be complicated by cardioembolism, heart failure, or ventricular arrhythmias, including sudden cardiac death (Finsterer, J. 2009).

REFERENCES